In Portugal, pediatric obesity is an important public health problem (30.7% children are overweight or obese).

In 95% of cases, obesity is exogenous (calory intake higher than energy output).

Genetic etiology should be considered in children with dysmorphic features, global developmental delay, early onset of severe obesity (before 5 years), hyperphagia or family history of severe obesity.

Regardless of etiology, the cornerstone of obesity treatment is the implementation of a healthy lifestyle.

**INTRODUCTION**

**CLÍNICAL CASE**

10 y.o. Pediatric Endocrinology due to Obesity and High stature

- Late pre-term delivery
- Weight and height AGA
- Normal development
- Atopic history
- No history of family obesity
- Mid-parental height = 165.5 cm (z-score + 0.36)
- Asymptomatic
- No hyperphagia
- Age of menarche 10.5 y
- Cushing-like signs
- Weight z-score +5.26
- Height z-score + 3.35
- BMI z-score +3.95
- MC4R gene heterozygous mutation
- GRHL gene heterozygous mutation
- AQP7 gene homozygous mutation
- MC4R variant inherited from father (not overweight)

**COMMENTS**

- The authors present an early-onset severe non-syndromic obesity case with mutations in three distinct genes.
- MC4R gene mutations are the main cause for genetic obesity. GHRL e AQP7 gene mutations have been described as obesity risk factors. However, the detected variants cannot individually explain this child obesity.
- The authors suggest a possible synergistic effect of all three mutations as the underlying cause.
- In children with early onset severe obesity, genetic etiology should always be considered.